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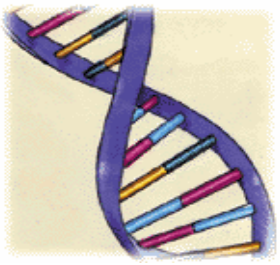
TRANSCRIPTIONS:

Genetics and Genomics in Public Health



Volume 1
Issue 3

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Genetic Disorders and Birth Defects Information Center

http://geneinfo.medlib.iupui.edu

By: E. Skopelja & K. Kaneshiro

Advances in human genetic research are rapidly changing how health professionals care for their patients. Genetic research impacts areas such as newborn screening, reproductive health, genetic testing, counseling and therapy. Patients and their families are being asked to make choices about such things as genetic testing and gene therapy in partnership with their health care providers. Public health professionals are incorporating genomics into large-scale strategies for disease detection, prevention, and treatment.

With all this research activity, health professionals need ready access to authoritative information in order to make informed decisions. A great deal of information from various sources does exist, especially on the Internet, but quality information can be difficult to locate, both for health care providers and patients.

Funded by a grant from the National Library of Medicine, our center has established a Web site devoted to untangling the mass of information on the Web. It is organized so that users can search for information by their population group, e.g. health professionals, or by topic, e.g. genetic testing and counseling. Information for health professionals includes links to sites on CME, statistics, directories, specialized databases, standards, practice guidelines, competencies, etc. **Topics** include anything related to genetic basics, genetic disorders, and birth defects. **Groups** include special populations that need a certain level or type of information related to their specific needs, e.g. Parents & Expectant Parents. Users can also type in keywords, such as “laboratories,” “clinics,” “sickle cell,” or “continuing education’ to quickly locate specific sites.

Most links have a brief annotation describing the content of the site, the source of the information, the audience level, the date, and other relevant facts. This should save users time by allowing them to decide whether the site is going to be useful before going there.

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Genetic Disorders and Birth Defects Information Center

Article by:

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&

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Indiana University School of Medicine Library

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It is also easy to direct patients to search for information on genetic or birth defect topics using the Center's Website. They will be linked to the most authoritative information on the Internet. If they cannot locate information on our site, they can call or email us with information requests. We will be glad to give them advice on searching the Internet or we will check our substantial collection of materials to locate an answer.

Health professionals can contact the center via phone, fax or email to check our large print collection of books and journals at the Ruth Lilly Medical Library. We can answer short questions, or refer people to additional resources if that is needed.

We hope that all types of health care providers and patients will utilize the center to make better informed decisions regarding this important topic. Please contact us with questions, comments or requests for fliers/posters at: **Genetic Disorders and Birth Defects Information Center** : http://geneinfo.medlib.iupui.edu

Focus on Family History:

National Coalition for Health Professional Education in Genetics 8th Annual Meeting

Article by:

Erin K. Herrick, Sc.M.

"Every child deserves a complete family medical history in their chart. It should be completed before they are born," according to Dr. Richard H. Carmona, the US Surgeon General and keynote speaker at National Coalition for Health Professional Education in Genetics (NCHPEG) 8th annual meeting. More than 200 attendees, including representatives from many of NCHPEG's 150 member organizations, and Krysta Barton on behalf of the Indiana State Department of Health, gathered for the meeting in Bethesda, Maryland, on January 27 and 28. Family history was the topic on everyone's mind, as was the take home message: A three-generation pedigree is the starting point for personalized, preventive medicine in the genomic era.

"As a surgeon, you can change one life a day, but in public health, you can change populations. We have to figure out how to move this country from a treatment-oriented society to a prevention-oriented society," said Dr. Carmona, a former surgeon who believes the key to success is health literacy. He challenged the audience to teach others about the importance of the family history in a culturally competent way, "so that you don't alienate the people you're trying to embrace."

The surgeon general launched his family history initiative and "My Family Health Portrait," a Web-based family history tool, on Thanksgiving day, 2004 – a time during the year when American families come together.

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The Risk of the Varicella-Zoster Virus Infection During Pregnancy

Jennifer Propst, M.S.; Paula R. Delk, M.S.; & David D. Weaver, M.D.

Varicella, also known as chickenpox, is caused by the varicella-zoster virus (VZV). Most often, the disease is acquired during childhood or immunity is conferred by vaccination. As a result, more than 95percent of women of childbearing age possess antibodies to the virus and are resistant to the disease. Because of lack of complete immunization to VZV, many cases of chickenpox/herpes zoster infections during pregnancy have been reported. The estimated incidence of varicella is 0.7 per 1,000 pregnancies or 3,000 annual US cases. In 25 percent of these cases, the maternal infection is transmitted to the fetus. Based on multiple prospective studies and case reports, maternal exposure and fetal infection during the fetus, has been found to be teratogenic; such an infection bears a moderate risk to the fetus. The possible fetal effects range from asymptomatic infection, to specific birth defects, to fetal demise. The constellation of features commonly found in affected fetuses is known as the fetal varicella syndrome (FVS). FVS has been observed in approximately 12 percent of infected fetuses and about 100 cases in infants have been reported. With the availability of the varicella-zoster immune globulin (VZIG) for prophylaxis and acyclovir as an antiviral treatment, a consensus is needed for counseling and treating women who are exposed to or infected with VZV. To assist in forming such a consensus, the following scenarios, ones commonly asked by health providers, are presented.

Q: A pregnant women who has no history of past chickenpox infection is exposed to an individual who has chickenpox. What should be done?
A: First, the woman should be tested for the presence of VZV antibodies. Many women will be found to be seropositive because of prior infection or having received VZV immunization. Reliable assays include an immunofluorescent assay (fluorescent anti-membrane antibody assay is preferred) or an enzyme-linked immunosorbent assay (ELISA). Recently, polymerase chain reaction (PCR) technique has been utilized for the detection of VZV in amniotic fluid and has been found reliable in detecting this infection. If the woman tests negative for VZV antibodies, administration of 500 units of varicella-zoster immune globulin (VZIG) is indicated when the exposure has occurred within 72-96 hours. Treatment with VZIG is primarily indicated to prevent varicella-related maternal pneumonia, which can have a mortality rate as high as 40 percent if untreated. In the fetus, VZIG treatment may not prevent varicella infection, but it may mitigate the effects of the infection. The fetal risk of the VZIG treatment appears to be small. For instance in one study (Enders et al, 1994), no cases of FVS or zoster infection in infancy occurred in 97 pregnancies in which maternal varicella infection developed following anti-varicella-zoster immune-globule prophylaxis. Only one infant from 89 of these pregnancies had elevated IgM antibody, a rate significantly lower than infected women who did not receive prophylaxis.

Q: A woman develops chickenpox during her pregnancy. What is the risk of FVS-related birth defects occurring in the fetus? How does the risk change by trimester?
A: Based on multiple prospective studies, the period of maximum risk to the fetus is highest when the maternal infection is contracted during the second trimester, or between 13-20 weeks gestation. The primary anomalies reported in FVS includes skin scarring, limb abnormalities, and brain and eye defects. Brain defects may include cerebral, cortical and cerebellar atrophy; nerve palsies, particularly Horner syndrome; and microcephaly. Seizures and developmental delay can also occur. Eye defects include cataracts, chorioretinitis, optic atrophy and microphthalmia. Gastrointestinal and genitourinary anomalies and fetal growth retardation have also been observed. Finally, approximately 25% of affected infants die in infancy.

When the mother contracts varicella infection in the first trimester, the risk of the fetus developing FVS is smaller (0.4-1 percent). After 20 weeks gestation, the risk for FVS is almost nonexistent; however, if a pregnant woman contracts VZV within three weeks prior to delivery, the newborn may develop overt chickenpox.

Extensive ultrasound is indicated at 16-22 weeks gestational age in women affected with VZV during the first half of pregnancy. Amniocentesis to evaluate for VZV in the amniotic fluid is only indicated if suspicious abnormalities are observed on ultrasound. Unfortunately, if the fetus is found to be infected, the severity of the disease and the pregnancy outcome cannot be predicted reliably.

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The Risk of the Varicella-Zoster Virus Infection During Pregnancy

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Q: A woman develops chickenpox anytime from three weeks before to three days after delivery. What are the risks to the fetus and what is the recommended treatment? Is acyclovir treatment for the mother and/or newborn appropriate?

A: The risk for symptomatic varicella infection in the infant is greatest when the mother contracts VZV six days prior to delivery to a few days after delivery. VZV infection in the neonate is of major concern since the infection if untreated, has a fatality rate of 30 percent. Neonatal disease is thought to occur due to transmission of the virus to the fetus but without transfer, and thus protection, of maternal VZV antibodies. Treatment of the prenatally exposed neonate with VZIG (125 units) is recommended. Isolation of an asymptomatic infant is also recommended from seven days after birth or from the onset of the maternal infection. Acyclovir treatment, either oral or intravenous, should be initiated in cases of severe neonatal infection.

It is not known whether acyclovir treatment of an infected mother during the first 20 weeks of pregnancy prevents FVS in the fetus, although there is evidence that doing so helps stop the progression of eye disease in the neonatal period. Although Acyclovir is not known to be teratogenic, there is an insufficient number of reported cases to state unequivocally that the drug is not teratogenic.

Q: Are children born to mothers who are affected with chickenpox during pregnancy at risk for developing other diseases?

A: Children born to mothers who had varicella during pregnancy are also at risk for developing herpes zoster, or shingles. This risk is the greatest when the mother developed VZV in the second half of pregnancy. These children also have an increased risk for developing cancer, especially leukemia, although the risk is only slightly greater than the general population risk. In one study, the risk of malignancy was 2 percent. Children born to women infected with VZV during pregnancy and who do not demonstrate any features of FVS typically have normal neurodevelopment.

Q: Is it safe for a woman infected with chickenpox to breastfeed?

A: VZV is not excreted in breast milk in an infected mother. In fact, the breast milk may contain antivaricella-zoster antibodies and may help protect the infant from infection. Therefore, breastfeeding is recommended, although attempts should be made to avoid direct contact of maternal lesions with the infant. Breast pumping and bottle feeding the breast milk would be an alternative to breastfeeding at these times.

Q: A woman received the chickenpox vaccine without knowing she was pregnant. What is the risk to the fetus?

A: The live attenuated VZV vaccine is not known to be teratogenic; however, there is insufficient data to state that there is no risk. The Advisory Committee on Immunization Practices of the U.S. Centers for Disease Control and Prevention (CDC) and the American Academy of Pediatrics recommend avoiding pregnancy for one month after receiving the vaccine. A registry (1-800-986-8999) has been established by Merck & Co., in collaboration with the CDC, to follow the outcomes of pregnancy in women who received the VZV vaccine during pregnancy. Of all the cases followed in the registry to date, no children born to women who had received the vaccine during pregnancy have had any FVS-associated birth defects.

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Focus on Family History

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The tool is available in English and Spanish at www.hhs.gov/familyhistory. Everyone is encouraged to use it to collect his or her family health history, and to share that information with health care provider(s).

Dr. Elizabeth Duke, senior administrator for the Health Resources and Services Administration, highlighted the "Healthy Choices Through Family History Awareness Project." This collaborative project brings together anthropologists, folklorists, genetic health specialists, and consumers to reach diverse African-American and Latino communities in Pennsylvania. The project relies on the oral tradition in those communities and focuses on storytelling, anecdotes, and other narratives used to transmit information and attitudes about health and disease. Ultimately, the project hopes to increase the community's genetics literacy and physicians' cultural literacy.

Meanwhile, the Centers for Disease Control and Prevention (CDC) is using the family history to target heart disease, stroke, diabetes, and breast, ovarian, and colorectal cancer – diseases that cause high rates of illness and death in the population at large. With the tacit admission that, "one size does not fit all," speakers Maren Scheuner, MD. MPH. and Paula Yoon, ScD. MPH. explained how the CDC proposes to use a family history tool to stratify risk and to tailor individual screening and preventive health messages.

Following a series of presentations on genetics-education programs for health professionals, Dr. Alan Guttmacher, deputy director of the National Human Genome Research Institute, provided some final thoughts on the subject of family history and its unique ability to capture not only individual genetic variation, but also shared elements that influence health status: the physical environment (e.g., sunlight and pollutants); the built environment (e.g., sidewalks, which promote walking and therefore better health); the dietary environment (e.g., meat, fat, junk food); the social environment (e.g., socioeconomic status and marital status); and the cultural environment (e.g. spirituality). As a tool for documenting the social, health, and environmental history of generations past, the future of the family history is assured. Look to the winter and spring issues of NCHPEG's Genetic Family History in Practice newsletter for more information and updates about these and other family history initiatives. In addition, speakers' PowerPoint presentations are available for free viewing and downloading at www.nchpeg.org. Visit the Website, or call NCHPEG at 410-583-0600 to learn more about the coalition's membership, its mission, and ongoing genetics education projects for health care professionals.

Fetal Alcohol Spectrum Disorder: Alcohol-Related Birth Defects that are 100% Preventable

On February 21, 2005, U.S. Surgeon General Richard H. Carmona warned pregnant women and women who may become pregnant to abstain from alcohol consumption in order to eliminate the chance of giving birth to a baby with any of the harmful effects of the Fetal Alcohol Spectrum Disorders (FASD). FASD is the full spectrum of birth defects caused by prenatal alcohol exposure.

Thirty-two years ago, U. S. researchers first recognized fetal alcohol syndrome (FAS). FAS is characterized by growth deficiencies (or decreased growth), specific facial features and central nervous system (or brain) abnormalities. The discovery of FAS led to considerable public education and awareness initiatives informing women to limit the amount of alcohol they consume while pregnant.

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But since that time, more has been learned about the effects of alcohol on a fetus. It is now clear that no amount of alcohol can be considered safe.

In the United States, FAS is the leading preventable birth defect with associated mental and behavioral impairment. There are many individuals exposed to prenatal alcohol who, while not exhibiting all of the characteristic features of FAS, do manifest lifelong neurocognitive and behavioral problems arising from this early alcohol exposure. In the United States, the prevalence of FAS is between 0.5 to 2 cases per 1,000 births. It is estimated that for every child born with FAS, three additional children are born who may not have the physical characteristics of FAS but still experience neurobehavioral deficits resulting from prenatal alcohol exposure that affect learning and behavior.

Recognizing the importance of prenatal alcohol exposure prevention, the **Indiana FASD Prevention task force** was formed last year as a subcommittee of the Indiana Genetics Advisory Committee (IGAC). The primary goal of the Indiana FASD Prevention Task Force is to implement a statewide FASD prevention campaign. In order to begin effective planning and implementation for a statewide FAS prevention campaign, evaluation of the current state of the State needs to be conducted to determine the incidence of alcohol use among women of childbearing age and to assess the knowledge of FAS and FASD among various target markets. The most efficient way to do this is by conducting a statewide needs assessment. Based on the results of that assessment, scheduled to be completed by March 31, 2006, a comprehensive strategic plan will be developed. The strategic plan will provide a guide for coordinating prevention activities across the state.

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